



HSD17B3 gene

hydroxysteroid 17-beta dehydrogenase 3

Normal Function

The *HSD17B3* gene provides instructions for making an enzyme called 17-beta hydroxysteroid dehydrogenase 3. This enzyme is active in the male gonads (testes), where it helps to produce the male sex hormone testosterone from a precursor hormone called androstenedione.

Health Conditions Related to Genetic Changes

17-beta hydroxysteroid dehydrogenase 3 deficiency

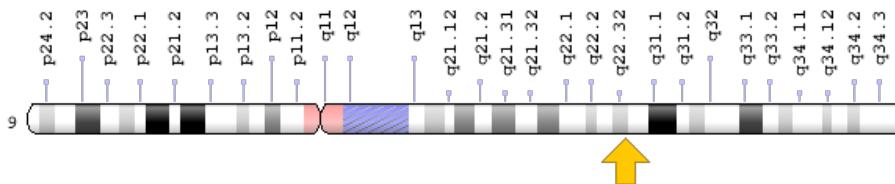
More than 20 mutations that cause 17-beta hydroxysteroid dehydrogenase 3 deficiency have been identified in the *HSD17B3* gene. In the Arab population of Gaza, where the condition is most common, almost all affected individuals have two copies of the same mutation. This mutation replaces the protein building block (amino acid) arginine with the amino acid glutamine at protein position 80 (written as Arg80Gln or R80Q).

Mutations in the *HSD17B3* gene result in a 17-beta hydroxysteroid dehydrogenase 3 enzyme with little or no activity, reducing testosterone production. A shortage of testosterone affects the development of the reproductive tract in the male fetus, resulting in the abnormalities in the external sex organs that occur in 17-beta hydroxysteroid dehydrogenase 3 deficiency.

Chromosomal Location

Cytogenetic Location: 9q22.32, which is the long (q) arm of chromosome 9 at position 22.32

Molecular Location: base pairs 96,235,306 to 96,313,987 on chromosome 9 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- 17-beta-HSD3
- DHB3_HUMAN
- EDH17B3
- estradiol 17 beta-dehydrogenase 3
- hydroxysteroid (17-beta) dehydrogenase 3
- SDR12C2

Additional Information & Resources

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28HSD17B3%5BTIAB%5D%29+OR+%28hydroxysteroid+++dehydrogenase+3%5BTIAB%5D%29%29+OR+%28%28estradiol+17+beta-dehydrogenase+3%5BTIAB%5D%29+OR+%2817-beta-HSD3%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- 17-BETA HYDROXYSTEROID DEHYDROGENASE III
<http://omim.org/entry/605573>

Research Resources

- **Atlas of Genetics and Cytogenetics in Oncology and Haematology**
http://atlasgeneticsoncology.org/Genes/GC_HSD17B3.html
- **ClinVar**
<https://www.ncbi.nlm.nih.gov/clinvar?term=HSD17B3%5Bgene%5D>
- **HGNC Gene Family: Short chain dehydrogenase/reductase superfamily**
<http://www.genenames.org/cgi-bin/genefamilies/set/743>
- **HGNC Gene Symbol Report**
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=5212
- **NCBI Gene**
<https://www.ncbi.nlm.nih.gov/gene/3293>
- **UniProt**
<http://www.uniprot.org/uniprot/P37058>

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